LABORATORY BULLETIN

DEPARTMENT OF HEALTH & ENVIRONMENTAL SCIENCES, HELENA, MONTANA

No. 56 David B. Lackman, Ph.D., Administrator, Laboratory Division, January 6, 1975

INFANT SCREENING TESTS FOR INBORN ERRORS OF METABOLISM

Today we joined the OREGON METABOLIC DISEASE SCREENING PROGRAM. This is the final stage in the implementation of Sections 69-6710 through 69-6713, R.C.M. 1947 and Montana Administrative Codes 16-2.18 (6) - S1820. For a copy of these regulations refer to Laboratory Bulletin No. 53, April 1, 1974. Under this program tests for phenylalanine, tyrosine, methionine (homocystinuria), leucine, galactosemia, and maple syrup urine disease will be done on each specimen. Others may be included on an experimental basis when indicated. Significant findings will be reported immediately to Steven Kairys, M.D., Chief, Maternal and Child Health Services Bureau, Montana State Department of Health and Environmental Sciences. Consultation will be available from the Maternal and Child Health Section of the Oregon Division of Health and the faculty of the Medical School.

Montana is fortunate in being invited to participate in this program because such a project is practical only when you are dealing with around 50,000 live births a year. We had 11,403 live births in the last fiscal year. The Oregon laboratory is automated and deals with a sufficient number of specimens to achieve adequate internal quality control. Some of these conditions have an incidence of less than 1:50,000. Alaska already is in the Oregon program and Idaho has it under consideration. For sparsely populated areas of the West, regional programs are a practical solution to some of our problems.

I will be spending Tuesday, January 21 in Portland with Rhesa L. Penn, Jr., M.D., Director, Maternal and Child Health Section and Gatlin R. Brandon, Director, Public Health Laboratory, Oregon State Health Division to discuss the program and to resolve any difficulties which may develop. I will also meet with the consultants at the medical school who may be involved with Montana physicians on their cases.

There are two controversial issues which have already been raised by material in Laboratory Bulletin No. 53, Infant Screening Tests. Although several persons have commented on them, the most authoritative criticisms have come from the Massachusetts Metabolic Disorders Screening Program, the pioneer in this field. The first question concerns the following section of our rules :

> (5) Required specimens for testing shall be taken by the hospital or institution wherein newborn care was rendered on the third day of life or 48 hours following ingestion of milk but not later than the 14th day of life.
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> (a) In the event the newborn is discharged from the hospital prior to the third day of life, the tests shall not be performed before discharge. In this case, it shall be the duty of the administrative officer or other person in charge of each hospital or institution caring for newborn infants to make provision at the time of discharge for the proper testing of the newborn and to explain the reasons why it is of utmost importance to return for these tests. The parent or legal guardian of the newborn shall also be required to sign a statement assuming responsibility to cause the tests to be administered between the third and 14th day of life.

(over)

Here is the reaction to this from the Massachusetts program. "I would like to venture the opinion that this is a terrible mistake and will almost surely result in a "missed" PKU baby at some future time. In Massachusetts we have always considered "a bird in hand is worth two in a bush" and in regard to PKU screening, this, we feel, is particularly important. As has been pointed out by Cunningham and his group in California, some PKU infants have increased blood phenylalanine levels even during the first 24 hours of life and before being fed. Our policy, therefore, is to insist the hospital send us a blood specimen on every baby at the time of nursery discharge, regardless of the age or feeding situation. We also insist that the baby who is discharged before the age of three days receive another blood test at the first "well-baby" visit."

In bulletin No. 53 I made some comments about screening of newborn for evidence of inherited hypothyroidism. These were in a somewhat negative vein so Massachusetts came through again with these remarks. "Furthermore, it is my impression that the cause of endemic cretinism in the United States is relatively unknown and, with the exception of those instances where the mother has been on antithyroid drugs or some other antithyroid agent, the etiological factors are not well defined. Consequently, the only reliable method of screening for hypothyroidism still involves the measurement of the serum thyroxin, serum triiodo-thyroxine and thyroid stimulating hormone. We at the State Laboratory are hoping to incorporate hypothyroid screening into the metabolic diseases program but, with the fiscal situation the way it is, it will probably be a long time before we can implement this."

For collection of specimens, a four-spot filter paper replaces the previous three-spot one. Please completely fill each ring as specified in instructions on the back of the form. Also write firmly so the information carries through to the carbon copies. Better yet, fill them out on the typewriter.

Item of information. An important document has recently been distributed by Comprehensive Health Planning, Department of Health and Environmental Sciences entitled "Montana State Plan for Health". Although it does not have a specific section dealing with laboratory services, there is much which is of concern to laboratories. It is available for reference in all public libraries, college and university libraries, and in the offices of the CHP Areawide organizations. Laboratory is specifically mentioned in the minutes of the meeting of July 26, 1974 at which this plan was approved, as follows :

> "Don Murray (M.D.) raised a concern about billing for laboratory services. In some cases physicians add on charges for lab services done outside their offices. Warren Croston stated that another matter which bothers him is when a doctor places a lab and X-ray in his office in competition with the hospital and he does not have to meet the licensure standards for these people that have to be met in the hospitals. The Personal Health Services and the Facilities committees will work together on these matters."